



## Case report

## Harlequin ichthyosis: A medico legal case report &amp; review of literature with peculiar findings in autopsy

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## ABSTRACT

Ichthyosis fetalis or Harlequin ichthyosis is an extremely severe and fatal hereditary skin disorder with an autosomal recessive inheritance. It is distinctive because of its remarkable clinical appearance which includes epidermal keratinization, hypoplasia of fingers or nails, malformation of ear and nose and incompatibility with life. This report describes a case of Harlequin ichthyosis along with detailed autopsy findings, which manifested the suspended animation in the early hours after delivery and showing a peculiar malformation of the cerebral hemispheres. To the best of the knowledge, this may be the first report of Harlequin ichthyosis in the forensic literature describing mal development of cerebral hemispheres and suspended animation in a case of Harlequin Ichthyosis.

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## 1. Introduction

Ichthyosis fetalis or Harlequin ichthyosis is an extremely severe and usually fatal hereditary skin disorder with an autosomal recessive inheritance.<sup>1</sup> It is a distinctive condition because of its remarkable clinical appearance and incompatibility with life. Its grotesque appearance includes large, thick greyish yellow armor-like skin plaques with reddish moist oozing cracks all over the body, severe bilateral ectropion, eclabium, flattened and malformed ears, malformed nose, and semi flexed extremities along with other features like absence of scalp hair, eyebrows and eyelashes.<sup>1</sup> This case report presents two peculiar associations to this condition.

## 2. Case report

A female baby was born at 39 weeks of intrauterine age in a government maternity hospital, without any signs of life at the time of birth. The baby was declared “Dead” and handed over to the parents by the doctor. At first the parents refused to take the body because of its grotesque appearance but later agreed after interference of the higher officials of the hospital. The parents made a contract with a lower grade employee of that hospital for disposal and funeral of the dead body of the fetus by offering some

monetary benefits to him. However, he discarded the so-called “Dead baby” in a dustbin very near to the hospital instead of burial. The fetus started crying and showed some movements and after 2 h and the local people reported this to the police. A medico legal case was registered under Sec. 317, and Sec. 318 of Indian Penal Code by the police and the live baby was admitted in perinatal ward. In spite of treatment the baby expired after 2 days and Medico-legal autopsy was conducted. The fetus had very thick dry, hard cracked grayish skin, with cracks having depth of 0.25–0.5 cms in some areas and the limbs were developed but the deformities were noted in hands and feet due to hyperkeratosis. (Fig. 1). There was ectropion of both eyes, eclabium of both lips and other deformities like sparse hair, rudimentary ears, and severe facial distortion (Fig. 2). The diagnosis of harlequin ichthyosis was made by pathognomonic features of the fetus. Apart from the typical findings, mal-development of both cerebral hemispheres (Fig. 3a) with normal skull bones and meninges were found during the autopsy. (Fig. 3b). Gross infective changes were found in both lungs.

## 3. Discussion

Harlequin ichthyosis is a rare and extremely severe form of congenital ichthyosis<sup>1</sup> with an incidence of about 1 in 300,000 births.<sup>2</sup> This condition is an autosomal recessive disease.<sup>3,4</sup> It is characterized by diamond shaped configuration of skin resembling the costume of a harlequin clown, severe ectropion and eclabium.<sup>4</sup> The harlequin ichthyosis has very bad prognosis as most of the

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**Fig. 1.** Harlequin fetus - showing very thick dry, hard cracked grayish skin, with 0.25–0.5 cms deep furrows in some areas, and the hands and feet showing deformities due to hyperkeratosis.

fetuses may die within few hours to few days after the birth from undernourishment caused by the rigidity of the lips, and respiratory distress due to under ventilation and pneumonia.<sup>2</sup>

Abnormal keratinocyte lamellar granules are a hallmark of Harlequin Ichthyosis skin.<sup>5</sup> An abnormal synthesis or metabolism of the lipid contents was previously suspected as being a possible pathogenic mechanism underlying Harlequin ichthyosis. Until 2005 causative gene ABCA 12 (Adenosine triphosphate binding cassette A12) had not been identified. Previously the prenatal diagnosis had been performed using electron microscopic examination of the fetal skin biopsy.<sup>6,7</sup> The discovery of the underlying gene causing Harlequin ichthyosis enabled DNA-based prenatal diagnosis of Harlequin ichthyosis by chorionic villus or amniotic fluid sampling in the earlier stages of pregnancy.<sup>7,8</sup> Severe Adenosine triphosphate binding cassette A12 (ABCA12) deficiency causes defective lipid transport via Lamellar Granule in keratinizing epidermal cells, resulting in the Harlequin Ichthyosis (HI) phenotype.<sup>5–7</sup> Adenosine triphosphate binding cassette A12 (ABCA12) is a keratinocyte lipid transporter associated with lamellar granule (LG) formation and lipid transport via lamellar granules (LG) on the surface of keratinocytes.<sup>6,7</sup> The characteristic



**Fig. 2.** Showing ectropion of both eyes and eclabium of the both the lips, sparse hair, rudimentary ears, and severe facial distortion.



**Fig. 3.** (a): Mal development of both cerebral hemispheres. (b): Development of skull bones normal. The photograph was taken after opening of the skull bones by the dissector during the autopsy.

clinical features of Harlequin Ichthyosis (HI) include thick, plate-like scales over the entire body with ectropion, eclabium and flattened ears.<sup>6,7</sup> Enlarged thymus was also reported in autopsy of Harlequin Ichthyosis.<sup>9</sup>

In the present case no abnormality was detected in thymus and the histopathology showed severe keratosis and acanthosis of the cells of the skin with diminution of elastic tissue. Absence of normal laminar neuronal arrangement and ischemic necrotic lesions were observed in brain and severe broncho pneumonic changes were observed in lungs and the cause of death was infection of both lungs in a case of harlequin ichthyosis.

The other finding in this case is suspended animation. The doctor who delivered the baby declared death, not completely realizing that the baby was in a state of suspended animation. This condition is most common in neonates and in violent deaths like electrocution and drowning and may also be possible voluntarily in practice of yoga. The incidence of this special condition in harlequin's fetus has not been reported in the literature available. The mal-development of brain in this baby has not been described in association with Harlequin Ichthyosis in literature available, and the frequency of this ill development of brain in harlequin fetus should be evaluated.

Though the case completely appeared as a natural death, the autopsy was done as a medico legal case as there is abandonment of

the fetus and fetal “dead body”( sec. 317, and 318 of Indian penal code<sup>10</sup>) Sec. 317 of Indian Penal Code<sup>10</sup> describes the punishment for Exposure and abandonment of child under twelve years, by parent or person having care of it and Sec. 318 of Indian Penal Code<sup>10</sup> describes that whoever, by secretly burying or otherwise disposing of the dead body of a child whether such child die before or after or during its birth shall be punished. In this case the police invoked the case under these two above mentioned sections of IPC on parents and the hospital worker who actually thrown the fetus in to dust bin. However it is a common practice in India for the police to invoke these two sections of IPC in cases of fetus found in dustbin due to ambiguity about the facts of the case. Much difficulty was faced to evaluate the legal issues during the trial.

#### 4. Conclusion

The death being inevitable in Harlequin ichthyosis cases, usually diagnostic pathologic autopsies will be performed, but in this instance a medico legal autopsy was conducted due to the circumstances of the case. Apart from all the classical clinical findings, the state of suspended animation and mal-development of the brain are the new findings in this harlequin's ichthyosis baby and the knowledge of all these findings in association with the state of suspended animation will help during autopsies in future.

#### Ethical approval

None.

#### Funding

None.

#### Conflict of interest

None.

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